
Case 4

Bony plaque on the leg

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Patient: A 22-year-old Thai woman from Lopburi

Chief complaint: Bony consistency plaques on left leg since the age of 7

Present illness:

The patient has developed a slowly progressive hard indurated plaque on her left leg since she was 7 years old. She denied previous major trauma to her left foot and leg. The plaques progressed over time, extended from left foot to below knee region. There was bony-like material extruded from the lesion when she scratched. She also had loose, lax skin over the hard plaques.

Past history:

- She was born at term following as uneventful pregnancy to nonconsanguineous parents.
- She has no underlying disease.

Family history:

No similar skin lesion among family members

Physical examination:

Height 160 c.m., Weight 52.5 kg.

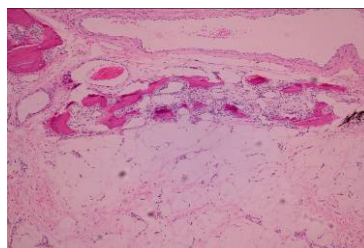
HEENT: No heliotrope, no alopecia, normal hearing

Cardiovascular and respiratory system: WNL

Abdomen: Soft, not tender, no hepatosplenomegaly

Central nervous system: WNL

Musculoskeletal: Mild limit ROM of left big toe, no sclerodactyly, no brachydactyly, no scoliosis and kyphosis



Skin examination:

- A large indurated skin-colored bone-like plaque with multiple hard brownish papulonodular surface extending from dorsal surface of left foot to lateral aspect of proximal part of left leg
- The overlying skin is loss of elasticity

Histopathology: (S14-17627, left leg)

- A proliferation of loose connective tissue composed of thin collagen bundles, scattered wavy small spindle cells with abundant mast cells
- Bone trabeculae composed of osteocytes and central fatty tissue among loose connective tissue

Investigation:Laboratory tests

Serum calcium, phosphate, parathyroid hormone: Normal

25-OH vitamin D level: 17.7 ng/ml (≥ 30 ng/ml)

GNAS1 gene mutation: Pending

Imaging studyFilm X-ray left foot and leg:

- Extensive patchy/plaque-like skin and soft tissue calcification along the distal left leg and foot
- Deformities of 3rd and 4th metatarsal bones left foot

Diagnosis: To be discussed

Treatment: To be discussed

Discussion:

Cutaneous ossification is a rare lesion of the skin encountered as both primary and secondary process. Primary cases are seen in the absence of any other preexisting lesion, whereas secondary cases are seen in relation to either neoplastic or inflammatory processes.¹

Primary cutaneous ossification have included entities such as progressive osseous heteroplasia (POH), fibrodysplasia ossificans progressiva (FOP), Albright hereditary osteodystrophy (AHO), plate-like osteoma cutis and miliary osteoma of the face as describes in table 1.²⁻⁵ Therefore careful evaluation of calcium-phosphate metabolism, combined with an assessment for associated systemic abnormalities, family history of unusual cutaneous ossification are necessary for the correct diagnosis.

Secondary cutaneous ossification have been reported in a variety of diseases, including pilomatricoma, basal cell carcinoma⁶, acne, melanocytic nevi, cellular blue nevus, scars, cutaneous mixed tumors, cylindromas, subcutaneous nodules of mixed connective tissue disease, dermatofibroma⁷, ossifying plexiform tumor⁸, pyogenic granuloma, cutis laxa-like pseudoxanthoma elasticum, organoid nevi, epidermal and dermoid cysts, and sites of trauma or injection.¹

The investigation for calcium-phosphate metabolism should be done in cutaneous ossification patient including serum levels of calcium, inorganic phosphate, alkaline phosphatase, parathyroid hormone (PTH) and vitamin D metabolites. Histopathology of cutaneous ossification shows well formed bony spicules with prominent cement lines and calcification. They may demonstrate

osteoblasts, osteoclasts, osteocytes and occasionally may even reveal bone marrow elements.

Our patient has developed progressive cutaneous ossification on her left foot and leg since she was 7 years old. She has neither anatomical nor significant laboratory abnormality. Histopathology of the lesion shows proliferation of loose connective tissue composed of thin collagen bundles, scattered wavy small spindle cells with abundant mast cells, bone trabeculae composed of osteocytes and central fatty tissue.

Currently, the results of GNAS1 gene mutation testing and special stains of the lesion (S 100, neurofilaments, factor 13a) are pending. Consequently the diagnosis for this patient will be discussed in the conference.

Table 1: The differential diagnosis of primary cutaneous ossification

	FOP	POH	AHO	Plate-like osteoma cutis	Miliary osteoma of the face
Mechanism of ossification	Endochondral	Intramembranous	Intramembranous	Intramembranous	Intramembranous
Clinical course	Progressive	Progressive	Limited	Limited	Limited
Associated feature	Malformed great toe	None	Obesity, brachydactyly, short stature, PTH resistance	None	None
Genetic mutation	ACVR1 gene	GNAS1 gene	GNAS1 gene	GNAS1 gene	Unknown
Mode of inherited	AD	AD	AD	AD	-

References:

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